Assessing the prevalence, impact, and clinical utility of genetic testing in children with cerebral palsy (CP) and physical neurodisabilites (PNDs)

Russell, K.,[1] Reefah, G.,[1] Rapley, J.,[1] Taylor, J.,[1,2] Yu, H.,[1] Peltekova, I.,[1] Kawamura, A.,[1,2] Baribeau, D.[1,2]
[1]Bloorview Research Institute [2]University of Toronto

Background and Rationale

- Meta-analyses suggest up to **31%** of children with CP have an identifiable rare genetic variant impacting development[1]
- Access to genetic testing for CP and/or PND is variable: guidance is needed regarding genetic testing strategies.[1]

Research Questions

1. What proportion of children with CP/PND underwent genetic testing?
2. What clinical factors were associated with a genetic diagnosis?
3. What was the clinical utility of the genetic diagnosis?

Design and Methods

Retrospective chart review of clients aged **≤18 years** seen in the Neuromotor Clinic at Holland Bloorview.

1. Cohort Identification (CP/PND) → 2. Data Extraction (i.e. clinical features) → 3. Clinical Utility Assessment

Of **260 children** with CP or a PND, **72%** received genetic testing, **55%** received a genetic diagnosis, of which **71%** had the potential to directly impact clinical care

Relevance to Holland Bloorview

Individualized Care Planning → Anticipatory Guidance → Family-Centered Care

Acknowledgements

Thank you to the Ward Family for funding this program. Thank you to the Neuromotor program clinicians, patients, and families. Thank you to the Kimmel Foundation for their contribution to this research.

Bloorview RESEARCH INSTITUTE

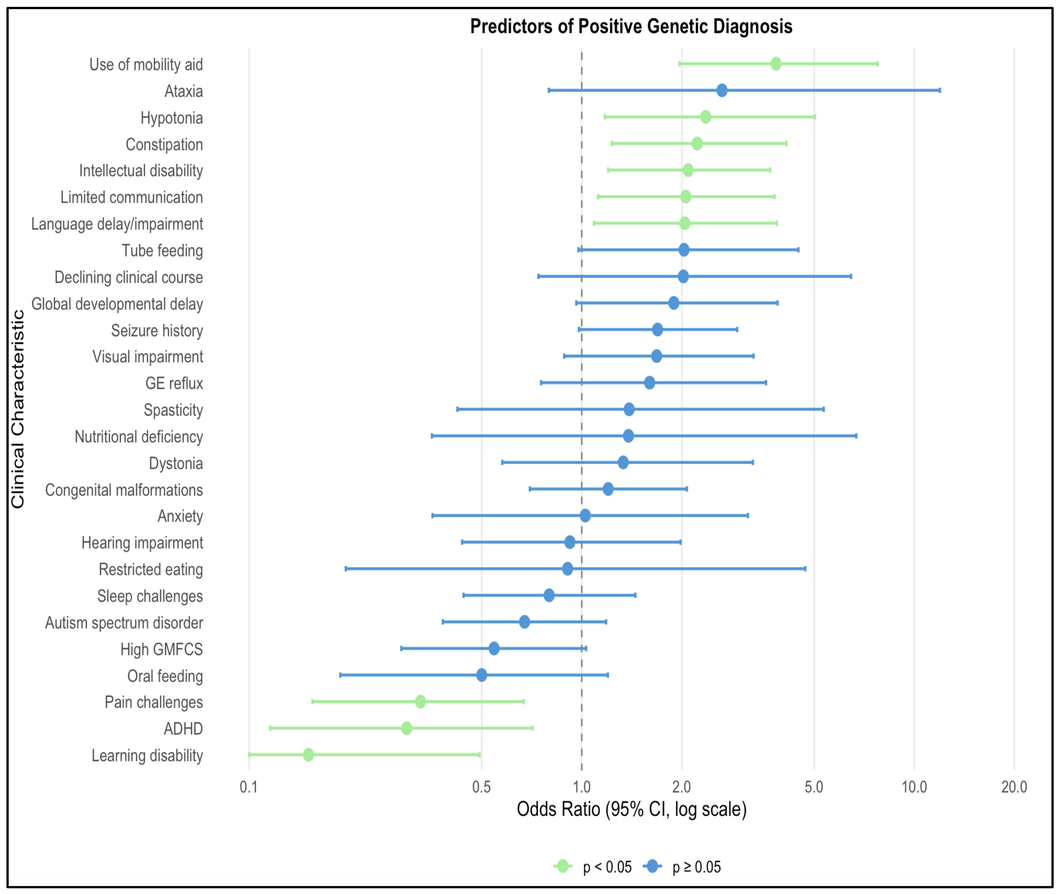
Holland Bloorview Kids Rehabilitation Hospital

Results

Aim 1: *What proportion of children with CP/PND underwent genetic testing?*

Of the 260 children diagnosed with either CP and/or a PND, **72% underwent genetic testing** (46% genome-wide sequencing).

Aim 2: *What clinical factors were associated with a genetic diagnosis?*



Aim 3: *What was the clinical utility of genetic diagnoses?*

A genetic diagnosis added actual or potential clinical utility to patient care for 71% of children. This included understanding the natural history/prognosis (66%), prompting additional referrals/investigations (64%), and/or choosing treatments (59%).

Conclusions and Next Steps

Genetic testing was common among children with CP/PNDs, and genetic diagnoses were identified in over half of those tested. These diagnoses can **add clinical utility** by informing investigations, prognosis, and care planning.

Next Steps: REB Amendment → Additional Variables (i.e. prematurity) → Future Analysis

1. Gonzalez-Mantilla, P.J., Hu, Y., Myers, S.M., et al. Diagnostic Yield of Exome Sequencing in Cerebral Palsy and Implications for Genetic Testing Guidelines: A Systematic Review and Meta-analysis. JAMA Pediatr. 2023;177(5):472–478. doi:10.1001/jamapediatrics.2023.0008